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# Hereditary cryohydrocytosis with reduced stomatin

INSERM

## Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Hereditary cryohydrocytosis with reduced stomatin. ORPHA:168577*

Hereditary cryohydrocytosis with reduced stomatin is a rare hemolytic anemia characterized by combination of neurologic features, such as psychomotor delay, seizures, variable movement disorders, and hemolytic anemia with stomatocytosis, resulting in cation-leaky erythrocytes, pseudohyperkalemia, hemolytic crises and hepatosplenomegaly. Cataracts are also a presenting feature.